

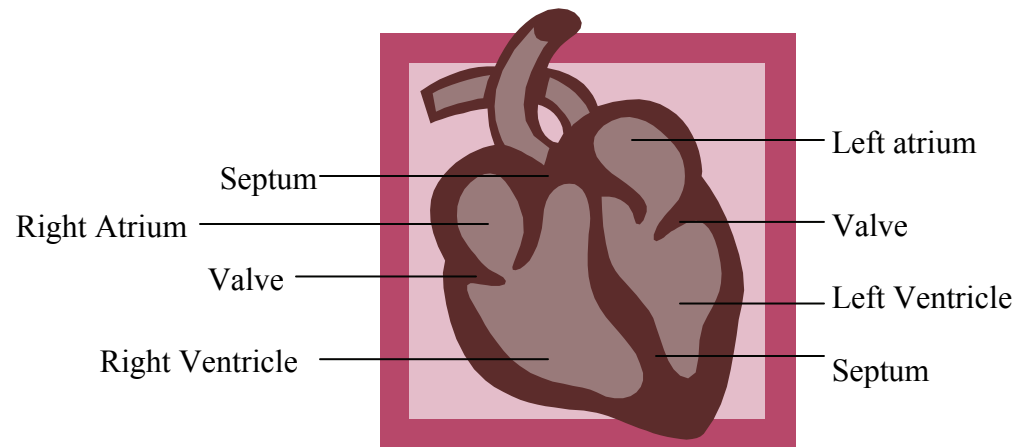
How is this study designed?

- Down syndrome is caused by an extra chromosome 21. The purpose of this study is to determine why some children born with Down syndrome have a heart defect, while others do not.
- CHD is the most common birth defect in people. Findings in this project will help us to understand CHD in all children.
- We will collect blood samples from children with Down syndrome with or without a heart defect and saliva samples from their parents. DNA will be isolated from the samples.
- We will compare DNA from families of children with a heart defect to DNA from families of children without a heart defect. By comparing the DNA we can identify genes that may play a role in the development of heart defects.
- We will use questionnaire information to identify environmental factors that influence a gene's ability to cause heart defects.
- We will make study results available to physicians, scientists, and all interested families.

What is a complete atrioventricular septal defect?

The human heart is divided into four chambers – the **left and right atria** and the **left and right ventricles** - by a combination of **septa** (partitions) and **valves**.

This heart structure ensures that blood entering the heart from the body is pumped to the lungs to pick up oxygen, returned to the heart, and pumped back to the body for oxygen delivery.



Efficient heart function is dependent upon the heart structures developing properly before birth.

An **atrioventricular septal defect (AVSD)** occurs when both septa and both valves do not develop properly. The resulting underdeveloped heart has difficulty pumping oxygen-carrying blood to the rest of the body. This heart defect requires surgical repair.

Who can participate in this study?

- Individuals with Down syndrome and a complete AVSD under the age of 18, and their parents
- Individuals with Down syndrome who were born with no heart defect and their parents may participate as control subjects

Participants in this study will be asked to:

- **Sign** a consent form
- **Provide** access to medical records such as a chromosome report and echocardiograms
- **Give** blood samples from the person with Down syndrome about 2 tablespoons from adolescents and about 1 teaspoon for smaller children. Children's blood samples may be taken at the time of routine blood work.
- **Give** saliva samples from the parents of the child with Down syndrome
- **Complete** telephone questionnaires (30-45 minutes) that covers health history, family health history, and environmental exposures.

Who is funding this study?

The study is funded by the National Institutes of Health (NIH).

How will you protect my privacy?

- Information you give us during the interview, as well as lab results from the samples you provide, will be kept confidential.
- Numbers instead of names will be used to identify participants' information.
- Personal information about you **will not** be available to anyone outside this study.
- We will **never** use your name or your family's name in any report or publication.

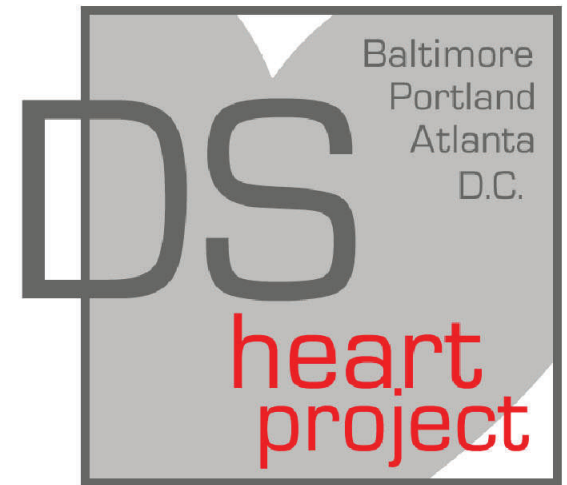
Contact Information

Kay Taylor
Kennedy Krieger Institute
Baltimore, Maryland
Research Coordinator II
443-923-9143 or 1-800-873-3377 ext. 9143
taylork@kennedykrieger.org

Katy Lesowski
Patient Recruitment Coordinator
Oregon Health & Science University
Portland, Oregon
503-494-7951
lesowski@ohsu.edu

Tracie Rosser, PhD
Research Project Coordinator
Emory University
Atlanta, Georgia
404-778-8474
trosser@emory.edu

Cindy Oxford-Wright, MS
Johns Hopkins University
Children's National Medical Center
Research Project Coordinator
678-559-6214



People with Down syndrome
helping to cure
congenital heart disease
in all people

- 40% of children born with Down syndrome (or trisomy 21) have a congenital heart defect (CHD).
- The most common form of CHD in Down syndrome is an atrioventricular septal defect (or AVSD)
- Children with Down syndrome are 2000 times more likely to have a complete AVSD than children without Down syndrome
- Researchers are conducting a multi-site study to identify genetic risk factors for congenital heart defects in people with Down syndrome.